

Application No. 09/109,082

K<sup>8</sup> (SEQ ID NO:10), including the amino acid terminus and the carboxy terminus. -

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IN THE CLAIMS:

Please replace claims 21, 30, 45, 52-54, 57, and 64-67 with the following amended claims.

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21. (Three times amended) A kit for the *in vitro* detection of a truncation, a deletion or a mutation in a survival motor neuron gene encoding the amino acid sequence of SEQ ID NO:22, comprising:

K<sup>9</sup> a pair of primers wherein said primers consist essentially of nucleic acid sequences selected from the group consisting of SEQ ID NOS:5-8 and 24-57;

reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

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K<sup>10</sup> 30. (Three times amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in a survival motor neuron gene encoding the amino acid sequence of SEQ ID NO:22, in a DNA sample, which comprises:

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- K<sup>10</sup>
- (a) amplifying said DNA in the sample with primers, wherein said primers consist essentially of nucleic acid sequences selected from the group consisting of SEQ ID NOS:5-8 and 24-57;
- (b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP) analysis, wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample to detect alterations in the patient gene; and
- (c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.
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45. (Amended) The method of claim 44, wherein said determining includes

K<sup>11</sup>

subjecting said exon 7 comprising nucleotide fragment to restriction enzyme digestion,

subjecting said exon 8 comprising nucleotide fragment to restriction enzyme digestion, and

analyzing enzymatic digestion products produced by said enzymatic digestions by comparing the enzymatic digestion products

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from the biological sample to enzymatic digestion products of exon 7 or exon 8 of the survival motor neuron gene from normal tissue,

K11 wherein an alteration of either exon 7 or exon 8 with reference to normal tissue is evidenced by an altered restriction enzymatic digestion pattern in one or both of said exons.

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52. (Amended) The method of claim 51, wherein said determining includes

subjecting said exon 7 comprising nucleotide fragment to restriction enzyme digestion,

subjecting said exon 8 comprising nucleotide fragment to restriction enzyme digestion, and

K12 analyzing enzymatic digestion products produced by said enzymatic digestions by comparing the enzymatic digestion products from the biological sample to enzymatic digestion products of exon 7 or exon 8 of the survival motor neuron gene from normal tissue,

wherein an alteration of either exon 7 or exon 8 with reference to normal tissue is evidenced by an altered restriction enzymatic digestion pattern in one or both of said exons.

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53. (Twice amended) A kit for the *in vitro* detection of a truncation, a deletion or a mutation in the Survival Motor Neuron gene of SEQ ID NO:22, wherein said kit comprises a probe which comprises at least 9 nucleotides within a sequence of SEQ ID NO: 12 or 13 or hybridizes with a sequence of SEQ ID NOS: 1, 2, or 10-13 under conditions having the stringency of 10% Dextran Sulphate Sodium, 1M NaCl, 0.05M Tris-HCl pH 7.5, 0.005M EDTA and 1% SDS at 65°C.

K13  
54. (Twice amended) A method of identifying the presence or absence of a mutation in the Survival Motor Neuron (SMN) gene of SEQ ID NO:22 in a nucleic acid sample, comprising

(a) subjecting the nucleic acid in the sample to digestion by a restriction endonuclease, wherein restriction fragments resulting from said digestion of a mutated SMN gene differ from those obtained from a T-BCD541 gene of SEQ ID NO:22 12; and

(b) identifying the presence or absence of a mutation in the SMN gene in the subject.

K14  
57. (Once Amended) The method of claim 56, wherein said polymerase chain reaction is performed with a set of primers

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K14 which are contained in the sequence comprising, or which comprise a sequence selected from SEQ ID Nos: 5 to 8 and 24 to 57.

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64. (Twice amended) A kit for the *in vitro* detection of a defect in the Survival Motor Neuron gene of SEQ ID NO:22, comprising:

a set of primers wherein said primers comprise a sequence selected from SEQ ID NOS: 5 to 8 and 24 to 57;

PCR reagents for amplifying DNA with said primers; and

a probe for the detection of the amplified product.

K15 65. (Twice amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene of SEQ ID NO:22, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers consisting essentially of nucleic acid sequences selected from the group of SEQ ID NOS: 5 to 8;

(b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP) analysis, wherein the analysis

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comprises comparing a pattern of DNA fragments obtained from the patient DNA sample to a pattern of DNA fragments obtained from a control DNA sample; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.

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66. (Amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene of SEQ ID NO:22, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers consisting essentially of nucleic acid sequences selected from the group of SEQ ID NOS: 24 to 57;

K16 (b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP), wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.

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67. (Amended) A method for detecting the presence or absence of a truncation, a deletion or a mutation in the Survival Motor Neuron gene of SEQ ID NO:22, wherein the gene is present in a DNA sample obtained from an individual, said method comprising :

(a) amplifying said DNA with primers, consisting essentially of nucleic acid sequences selected from the group of sequences which are inverted complementary sequences to SEQ ID NOS: 5 to 8;

klb  
(b) subjecting said amplified DNA to a Single-Strand Conformation Polymorphism (SSCP), wherein the analysis comprises comparing a pattern of DNA fragments obtained from the patient sample to a pattern of DNA fragments obtained from a control sample; and

(c) detecting the presence or absence of said truncation, deletion or mutation in the Survival Motor Neuron gene.

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